Dear Editor:

We are very pleased to learn from your letter about revision for our manuscript [Manuscript ID: 54243, "Early-Onset Refractory Diarrhea of Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-Linked (IPEX) Syndrome Associated with a Novel Mutation in the FOXP3 Gene: A case report"]. Thank you for your attention and the reviewers for their helpful comments. We have revised the manuscript according to the comments from the reviewers. In addition, the figures of editable are in the PowerPoint document. Below are our responses to the reviewer's comments.

Yours sincerely,

Riling Chen

Responds to the reviewer's comments:

1. #How many mutations have been reported so far about IPEX syndrome? The presently discovered mutation is the 3rd, or 4th , or 10th, or whatever? Please make a table showing every mutation of IPEX syndrome reported so far, including the present one.

Response: 99 mutations have been reported so far about IPEX syndrome (**Table 1**). This report mutation is the 100th.

2. There are a few careless grammatical mistakes: ex. Figure 1, figure legend: the point inside the circle indicate> the point inside the circle indicates.

Response: Thank you for pointing out my mistake. The "the point inside the circle indicate" in figure legend of the Figure 1 had be revised "the point inside the circle indicates". Revised portion are marked in red in the manuscript.

Table1 *FOXOP3* mutations in reported IPEX patients

Number	Domain	Exon or Intron	Mutation	Amino acid change	References
1	Non-coding	Intron −1	g.(−)6247_(−)4859del	NA	Gavin et al, 2006[1]; Torgerson et al, 2007[2]; Moes et al, 2010[3]
2	Non-coding	Intron −1	g.−1121T>G	NA	Moes et al, 2010[3]
3	Non-coding	Intron −1	c.−23G>A	NA	Sheikine et al, 2015[4]; Bis et al, 2015[5]
4	Non-coding	Intron −1	c.−23+1G>T	NA	Kadakia et al, 2019[6]
5	Non-coding	Intron −1	c.1_23G>T	NA	Kobayashi et al, 2011[7]
6	Non-coding	Intron 1	c.1_7G>T	NA	Myers et al, 2006[8]
7	PRR	Exon 1	c.2T>C	p.M1T	Gambineri et al, 2008[9]
8	PRR	Exon 1	c.3G>A	p.M1I	Bacchetta et al, 2006[10]; Gambineri et al, 2008[9]
9	PRR	Exon 1	c.151C > T	p.R51X	Shanes et al, 2019[11]
10	PRR	Exon 1	c.200G>T	p.Q70H	Heltzer et al, 2007[12]; Yong et al, 2008[13]
	Non-coding	Intron 1	c.201+1G>A	NA	Bae et al, 2011[14]
11	Non-coding	Intron 1	c.210_210 +1delGG>AC	NA	Gavin et al, 2006[1]; d'Hennezel et al, 2012[15]
12	Non-coding	Intron 1	c.210+1G>A	NA	Tsuda et al, 2010[16]
13	Non-coding	Intron 1	c.210+1G>T	NA	Otsubo et al, 2011[17]; Lin et al, 2018[18]
14	Non-coding	Intron 1	c.210+1G>C	NA	Fontenot et al, 2003[19]; Passerini et al, 2019[20]
15	Non-coding	Intron 1	c.210+2T>G	NA	Gambineri et al, 2008[9]; Passerini et al, 2011[21]
16	Non-coding	Intron 1	c.210+2delT	NA	Burroughs et al, 2010[22]
17	PRR	Exon 2	c.227delT	p.L76Qfsx53	Kobayashi et al, 2001[23]; Owen et al, 2003[24]; Fuchizawa et al, 2007[25]; Rubio-Cabezas et al, 2009[26]; Otsubo et al, 2011[17];

					Kobayashi et al, 2011[7]; Duclaux-Loras et al, 2015[27]
18	PRR	Exon 2	c.303_304delTT	p.F102Hfs*103	Rao et al, 2007[28]; Moudgil et al, 2007[29]
19	PRR	Exon 3	c.319_320delTC	p.Ser107/Asnfs*204	Xavier-da-Silva et al, 2015[30]
20	PRR	Exon 3	c.323C>T	p.T108M	De Benedetti et al, 2006[31]
21	PRR	Exon 3	c.398C>T	p.P133L	Wang et al, 2018[32]
22	Non-coding	Intron3	c.454+4A>G	NA	De Benedetti et al, 2006[31]
23	PRR	Exon 5	c.542G>A	p.S181A	This report
24	PRR	Exon 5	c.543C>T	p.S181S	Wildin et al, 2002[33]; Gambineri et al, 2008[9]; Zennaro et al, 2012[34]
25	PRR	Exon 5	c.560C>T	p.F187L	Halabi-Tawil et al, 2009[35]
26	PRR	Exon 5	c.560C>T	p.P187L	Patey-Mariaud de Serre et al, 2009[36]; Moes et al, 2010[3]
27	ZF	Exon 5	c.600delGGA	p.ΔE201	Chatila et al, 2000[37]
28	Non-coding	Exon 6	c.694T>G	p.C232G	Okou et al, 2014[38]
29	LZ	Exon 6	c.725T>C	p.L242P	Gambineri et al, 2008[9]; Passerini et al, 2011[21]; Baris et al, 2014[39]
30	LZ	Exon 6	c.727delG	p.Glu243Serfs*11	Louie et al, 2017[40]
31	LZ	Intron 6	c.736-1G>A	p.L246_V272del	Halabi-Tawil et al, 2009[35]
32	LZ	Intron 6	c.736-2A>C	NA	Chen et al, 2016[41]
33	LZ	Intron 6	c.736-2A>G	NA	Park et al, 2015[42]
34	LZ	Exon 7	c.748_750delAAG	p.K250del	Wildin et al, 2002[33]; Lopes et al, 2006[43]; Hashimura et al, 2009[44]; Otsubo et al, 2011[17]
35	LZ	Exon 7	c.748_750delAAG	p.ΔL250	Wildin et al, 2002[33]; Li et al, 2007[45]; d'Hennezel et al, 2012[15]; Luo et al, 2018[46]
36	LZ	Exon 7	c.749delA	p.Lys250Argfs*4	Louie et al, 2017[40]
37	LZ	Exon 7	c.750-752delGGA	p.Glu251del	Gavin et al, 2006[1]; Moes et al, 2010[3]; Baris et al, 2014[39]; Dong et al, 2018[47]
38	LZ	Exon 7	c.751_753del	p.Glu251del	Rodrigo et al, 2013[48]
39	LZ	Exon 7	c.751_753delGAG	P.E251del	Halabi-Tawil et al, 2009[35]; Patey-Mariaud de Serre et al,

					2009[36]; Moes et al, 2010[3]
40	LZ	Exon 7	c.751_753delCAG	p.E251del	Sheikine et al, 2015[4]
41	LZ	Exon 7	c.758T>C	p.L253P	Nademi et al, 2014[49]
42	LZ	Exon 7	c.767T>C	p.M256T	Dong et al, 2018[47]
43	Non-coding	Intron 7	c.816+2delT	NA	Passerini et al, 2011[21]
44	Non-coding	Intron 7	c.816+4A>G	NA	Harbuz et al, 2010[50]
45	Non-coding	Intron 7	c.816+5G>A	NA	Gambineri et al, 2008[9]; Baris et al, 2014[39]; Nademi et al, 2014[49]
46	LZ	Intron 7	c.816+7G>C	NA	Burroughs et al, 2010[22]; Tsuda et al, 2010[16]; Harbuz et al, 2010[50]
47	Non-coding	Intron 7	c.817-1G>A	NA	Heltzer et al, 2007[12]
48	LZ-FKH loop	Exon 8	c.817G>T	p.A273S	Tsuda et al, 2010[16]
49	LZ-FKH loop	Exon 8	c.876A>G	-	Nademi et al, 2014[49]
50	LZ-FKH loop	Exon 8	c.906delT	p.D303fs*87	Shehab et al, 2017[51]
51	LZ-FKH loop	Exon 8	c.935G>A	p.R312H	Wang et al, 2018[32]
52	Non-coding	Intron 8	c.967+3A>T	NA	Zhu et al, 2018[52]
53	Non-coding	Intron 8	c.967+4A>G	NA	Chatila et al, 2000[37]; Gambineri et al, 2008[9]; Passerini et al, 2011[21]
54	Non-coding	Intron 8	c.968-20A>C	NA	Bindl et al, 2005[53]
55	Non-coding	Intron 8	c.968+4A>G	NA	Gambineri et al, 2008[9]
56	LZ-FKH loop	Exon 9	g.11628T>C	p.F324L	An et al, 2011[54]
57	LZ-FKH loop	Exon 9	c.970T>C	p.F324L	Bacchetta et al, 2006[10]; Gambineri et al, 2008[9]; McMurchy et al, 2010[55]
58	LZ-FKH loop	Exon 9	c.972C>T	p.F324L	Bacchetta et al, 2006[10]; Gambineri et al, 2008[9]; d'Hennezel et al, 2012[15]
59	LZ-FKH loop	Exon 9	c.1009C>T	p.R337X	Reichert et al, 2015[56]

60	LZ-FKH loop	Exon 9	c.1009C>T	p.A337T	Rae et al, 2015[57]
61	FKH	Exon 9	c.1010G>A	p.R337Q	Rubio-Cabezas et al, 2009[26]; Savova et al, 2014[58]; Sheikine et al, 2015[4];Tuijnenburg et al, 2017[59]; Hoshino et al, 2019[60]
62	FKH	Exon 9	c.1010G>A	p.A337G	Savova et al, 2014[58]
63	FKH	Exon 9	c.1015C>T	p.P339S	Agakidis et al, 2019[61]
64	FKH	Exon 9	c.1015C>G	p.P339A	Gambineri et al, 2008[9]; Rubio-Cabezas et al, 2009[26]; Passerini et al, 2011[21]; Moes et al, 2010[3]
65	FKH	Exon 9	c.1033C>T	p.L345P	Vasiljevic et al, 2015[62]; Shehab et al, 2017[51]
66	FKH	Exon 9	c.1037T>C	p.I346T	Passerini et al, 2011[21]; Nademi et al, 2014[49]
67	FKH	Exon 9	c.1040G>A	p.R347H	Wildin et al, 2002[33]; Gambineri et al, 2008[9]; Scaillon et al, 2009[63]; McMurchy et al, 2010[55]; Passerini et al, 2011[21]; Seidel et al, 2016[64]; Hoshino et al, 2019[60]; Yamauchi et al, 2019[65]
68	Non-coding	Intron 9	c.1044+4A>G	p.E323Gfs X14	Chatila et al, 2000[37]; d'Hennezel et al, 2012[15]
69	Non-coding	Intron 9	c.1044+459A>G	NA	Wildin et al, 2002[33]
70	Non-coding	Intron 9	c.1045-3C> G	NA	Costa-Carvalho et al, 2008[66]
71	FKH	Exon 10	c.1061delC	p.P354Q	Heltzer et al, 2007[12]; Yong et al, 2008[13]
72	FKH	Exon 10	c.1080-1081insA	p.N361Kfs X2	An et al, 2011[54]; d'Hennezel et al, 2012[15]
73	FKH	Exon 10	c.1087A>G	p.I363V	Kobayashi et al, 2001[23]; Lopes et al, 2006[43]; McMurchy et al, 2010[55]
74	FKH	Exon 10	c.1099T>C	p.F367L	Suzuki et al, 2007[67]; Colobran et al, 2016[68]
75	FKH	Exon 10	c.1099 T>G	p.P367V	Colobran et al, 2016[68]
76	FKH	Exon 10	c.1100T>G	p.F367C	Patey-Mariaud de Serre et al, 2009[36]
77	FKH	Exon 10	c.1101C>G	p.F367L	Suzuki et al, 2007[67]; Halabi-Tawil et al, 2009[35]

78	FKH	Exon 10	c.1110G>A	p.M370I	An et al, 2011[54]
79	FKH	Exon 10	c.1113T> G	p.F371C	Wildin et al, 2001[69]; Baud et al, 2001[70]; Patey-Mariaud de Serre et al, 2009[36]
80	FKH	Exon 10	c.1117T>G	p.F373V	Tanaka et al, 2005[71]; Fuchizawa et al, 2007[25]; d'Hennezel et al, 2012[15]
81	FKH	Exon 10	c.1117T>G	p.P373V	Fuchizawa et al, 2007[25]; Otsubo et al, 2011[17]
82	FKH	Exon 10	c.1117-1118delTT> GC	p.F373A	Bacchetta et al, 2006[10]; Fuchizawa et al, 2007[25]; Gambineri et al, 2008[9]; McMurchy et al, 2010[55]; Passerini et al, 2011[21]
83	FKH	Exon 10	c.1121T>G	p.F374C	Halabi-Tawil et al, 2009[35]; Patey-Mariaud de Serre et al, 2009[36]; Gambineri et al, 2008[9]; Moes et al, 2010[3]
84	FKH	Exon 10	c.1139C>T	p.T380I	Zhan et al, 2008[72]
85	FKH	Exon 11	c.1150G>A	p.A384T	Bennett et al, 2001[73]; Wildin et al, 2001[69]; Nieves et al, 2004[74]; Taddio et al, 2007[75]; Gambineri et al, 2008[9]; Kasow et al, 2011[76]; Otsubo et al, 2011[17]; d'Hennezel, Eva et al, 2012[15]; He et al, 2017[77]; Griswold et al, 2018[78]; Zhu et al, 2018[52]; Hoshino et al，2019[60]; Bachelerie et al, 2019[79]
86	FKH	Exon 11	c.1157G>A	p.R386H	Tsuda et al, 2010[16]; Nademi et al, 2014[49]
87	FKH	Exon 11	c.1169G>A	p.S390N	Myers et al, 2006[8]; Tsuda et al, 2010[16]
88	FKH	Exon 11	c.1189C>T	p.R397W	Levy-Lahad et al, 2001[80]; Wildin et al, 2001[69]; Banerjee-Basu et al, 2004[81]; Lopes JE et al, 2006[43]
89	FKH	Exon 11	c.1189C>T	p.A397T	Xavier-da-Silva et al, 2015[30]
90	FKH	Exon 11	c.1190G>A	p.A397Q	Tsuda et al, 2010[16]; Barzaghi et al, 2018[82]
91	FKH	Exon 11	c.1190G>A	p.R397Q	Tsuda et al, 2010[16]; Ge et al, 2017[83]

92	FKH	Exon 11	c.1222G>A	p.V408M	Rubio-Cabezas et al, 2009[26]; Hashimura et al, 2009[44]; Wang et al, 2018[32]
93	FKH	Exon 11	c.1226A>G	p.D409G	Rao et al, 2007[28]
94	FKH	Exon 11	c.1227_1235del	p.A409G	Smith et al, 2016[84]
95	FKH	Exon 11	c.1227_1235del	p.D409_L411del	Alkorta-Aranburu et al, 2014[85]
96	C-terminal	Exon 11	c.1271G>A	p.C424Y	Lopes et al, 2006[43]; Rao et al, 2007[28]; Burroughs et al, 2007 [86]
97	Stop codon	Exon 11	c.1290-1309del-insTGG	p.G430fsX22	Peake et al, 1996[87]; Wildin et al, 2001[69]; d'Hennezel et al, 2012[15]
98	Stop codon	Exon 11	c.1293_1294delCT	ter432TfsX26	Bennett et al, 2001[73]; Wildin et al, 2002[33]; Kobayashi et al, 2011[7]
99	PolyA	PolyA	c.***876A>G (AAUAAA>AAUGAA)		Bennett et al, 2001[88]; Tsuda et al, 2010[16]; Nademi et al, 2014[49]
100	PolyA	PolyA	c.***878A>G (AAUAAA>AAUAAG)		Dorsey et al, 2009[89]

NA, not available; PRR, N-terminal proline-rich domain; ZF, zinc-fifinger domain; LZ, leucine-zipper domain; FKH, forkhead domain.

References

1. **Gavin MA**, Torgerson TR, Houston E, DeRoos P, Ho WY, Stray-Pedersen A, Ocheltree EL, Greenberg PD, Ochs HD. Single-cell analysis of normal and FOXP3-mutant human T cells: FOXP3 expression without regulatory T cell development. *Proc Natl Acad Sci U S A* 2006;**103**: 6659-6664. [PMID:16617117 DOI:10.1073/pnas.0509484103]
2. **Torgerson TR,** Linane A, Moes N, Anover S, Mateo V, Rieux-Laucat F, Hermine O, Vijay S, Gambineri E, Cerf-Bensussan N, Fischer A, Ochs HD, Goulet O, Ruemmele FM. Severe food allergy as a variant of IPEX syndrome caused by a deletion in a noncoding region of the FOXP3

gene. *Gastroenterology* 2007;**132**: 1705-1717. [PMID:17484868 DOI:10.1053/j.gastro.2007.02.044]

3. **Moes N,** Rieux–Laucat F, Begue B, Verdier J, Neven B, Patey N, Torgerson TT, Picard C, Stolzenberg MC, Ruemmele C, Rings EH, Casanova JL, Piloquet H, Breton A, Ochs HD, Hermine O, Fischer A, Goulet O, Cerf-Bensussan N, Ruemmele FM. Reduced expression of FOXP3 and regulatory T-cell function in severe forms of early-onset autoimmune enteropathy. *Gastroenterology* 2010;**139**: 770-778. [PMID:20537998 DOI:10.1053/j.gastro.2010.06.006]
4. **Sheikine Y,** Woda CB, Lee PY, Chatila TA, Keles S, Charbonnier L-M, Schmidt B, Rosen S, Rodig NM. Renal involvement in the immunodysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) disorder. *Pediatr Nephrol* 2015;**30**: 1197-1202. [PMID:25911531 DOI:10.1007/s00467-015-3102-x]
5. **Bis S,** Maguiness SM, Gellis SE, Schneider LC, Lee PY, Notarangelo LD, Keles S, Chatila TA, Schmidt BA, Miller DD. Immune dysregulation, polyendocrinopathy, enteropathy, X‐linked syndrome associated with neonatal epidermolysis bullosa acquisita. *Pediatr Dermatol* 2015;**32**: e74-e77. [PMID:25790289 DOI:10.1111/pde.12550]
6. **Kadakia S,** Farnaes L, Dimmock D, Chowdhury S, Ding Y, Anderson EJ, Kingsmore S, Newfield RS. Diagnosis and treatment of a boy with IPEX syndrome presenting with diabetes in early infancy. *Clin Case Rep* 2019;**7**: 2123-2127. [PMID:31788263 DOI:10.1002/ccr3.2438]
7. **Kobayashi I,** Kubota M, Yamada M, Tanaka H, Itoh S, Sasahara Y, Whitesell L, Ariga T. Autoantibodies to villin occur frequently in IPEX, a severe immune dysregulation, syndrome caused by mutation of FOXP3. *Clin Immunol* 2011;**141**: 83-89. [PMID:21741320 DOI:10.1016/j.clim.2011.05.010]

8. **Myers A,** Perroni L, Costigan C, Reardon W. Clinical and molecular findings in IPEX syndrome. *Arch Dis Child* 2006;**91**: 63-64. [PMID:16371377 DOI:10.1136/adc.2005.078287]

9. **Gambineri E,** Perroni L, Passerini L, Bianchi L, Doglioni C, Meschi F, Bonfanti R, Sznajer Y, Tommasini A, Lawitschka A, Junker A, Dunstheimer D, Heidemann PH, Cazzola G, Cipolli M, Friedrich W, Janic D, Azzi N, Richmond E, Vignola S, Barabino A, Chiumello G, Azzari C, Roncarolo MG, Bacchetta R. Clinical and molecular profile of a new series of patients with immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome: inconsistent correlation between forkhead box protein 3 expression and disease severity. *J Allergy Clin Immunol* 2008;**122**: 1105-1112. e1. [PMID:18951619 DOI:10.1016/j.jaci.2008.09.027]

10. **Bacchetta R,** Passerini L, Gambineri E, Dai M, Allan SE, Perroni L, Dagna-Bricarelli F, Sartirana C, Matthes-Martin S, Lawitschka A, Azzari C, Ziegler SF, Levings MK, Roncarolo MG. Defective regulatory and effector T cell functions in patients with FOXP3 mutations. *J Clin Invest* 2006;**116**: 1713-1722. [PMID:16741580 DOI:10.1172/JCI25112]

11. **Shanes E,** Propst L, Ouyang DW, Ernst LM. Recurrent Non Immune Fetal Hydrops Associated With IPEX Syndrome. *Pediatr Dev Pathol* 2019;**22**: 465-471. [PMID:30813833 DOI:10.1177/1093526619834809]

12. **Heltzer ML,** Choi JK, Ochs HD, Sullivan KE, Torgerson TR, Ernst LM. A potential screening tool for IPEX syndrome. *Pediatr Dev Pathol* 2007;**10**: 98-105. [PMID:17378693 DOI:10.2350/06-07-0130.1]

13. **Yong PL,** Russo P, Sullivan KE. Use of sirolimus in IPEX and IPEX-like children. *J Clin Immunol* 2008;**28**: 581. [PMID:18481161 DOI:10.1007/s10875-008-9196-1]

14. **Bae KW,** Kim BE, Choi J-H, Lee JH, Park YS, Kim G-H, Yoo HW, Seo JJ. A novel mutation and unusual clinical features in a patient with immune dysregulation, polyendocrinopathy, enteropathy, X-linked

(IPEX) syndrome. *Eur J Pediatr* 2011;**170**: 1611-1615. [PMID:21979562 DOI:10.1007/s00431-011-1588-1]

15. **d'Hennezel E,** Dhuban KB, Torgerson T, Piccirillo CA. The immunogenetics of immune dysregulation, polyendocrinopathy, enteropathy, X linked (IPEX) syndrome. *J Med Genet* 2012;**49**: 291-302. [PMID:22581967 DOI:10.1136/jmedgenet-2012-100759]
16. **Tsuda M,** Torgerson TR, Selmi C, Gambineri E, Carneiro-Sampaio M, Mannurita SC, Leung PS, Norman GL, Gershwin ME. The spectrum of autoantibodies in IPEX syndrome is broad and includes anti-mitochondrial autoantibodies. *J Autoimmun* 2010;**35**: 265-268. [PMID:20650610 DOI:10.1016/j.jaut.2010.06.017]
17. **Otsubo K,** Kanegane H, Kamachi Y, Kobayashi I, Tsuge I, Imaizumi M, Sasahara Y, Hayakawa A, Nozu K, Iijima K, Shuichi I, Reiko H, Yoshinon N, Kiyoshi T, Hisashi M, Hans DO, Toshio M. Identification of FOXP3-negative regulatory T-like (CD4+ CD25+ CD127low) cells in patients with immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. *Clin Immunol* 2011;**141**: 111-120. [PMID:21802372 DOI:10.1016/j.clim.2011.06.006]
18. **Lin Y,** Xu A, Zeng C, Cheng J, Li N, Niu H, Liu L, Li X. Somatic and germline FOXP3 mosaicism in the mother of a boy with IPEX syndrome. *Eur J Immunol* 2018;**48**: 885-887. [PMID:29400909 DOI:10.1002/eji.201747445]
19. **Fontenot JD,** Gavin MA, Rudensky AY. Foxp3 programs the development and function of CD4+ CD25+ regulatory T cells. *Nat Immunol* 2003;**4**: 330-336. [PMID:12612578 DOI:10.1038/ni904]
20. **Passerini L,** Barzaghi F, Curto R, Sartirana C, Barera G, Tucci F, Albarello L, Mariani A, AlbertoTestoni P, Bazzigaluppi E, Bosi E, Lampasona V, Neth O, Zama D, Hoenig M, Schulz A, Seidel MG, Rabbone I, Olek S, Roncarolo MG, Cicalese MP, Aiuti A, Bacchetta R. Treatment with Rapamycin Can Restore Regulatory T Cell Function in

IPEX Patients. *J Allergy Clin Immunol* 2019. [PMID:31874182 DOI:10.1016/j.jaci.2019.11.043]

21. **Passerini L,** Olek S, Di Nunzio S, Barzaghi F, Hambleton S, Abinun M, Tommasini A, Vignola S, Cipolli M, Amendola M, Naldini L, Guidi L, Cecconi M, Roncarolo,MG, Bacchetta R. Forkhead box protein 3 (FOXP3) mutations lead to increased TH17 cell numbers and regulatory T-cell instability. *J Allergy Clin Immunol* 2011;**128**: 1376-1379. e1. [PMID:22000569 DOI:10.1016/j.jaci.2011.09.010]

22. **Burroughs LM,** Torgerson TR, Storb R, Carpenter PA, Rawlings DJ, Sanders J, Scharenberg AM, Skoda-Smith S, Englund J, Ochs HD, Woolfrey AE. Stable hematopoietic cell engraftment after low-intensity nonmyeloablative conditioning in patients with immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. *J Allergy Clin Immunol* 2010;**126**: 1000-1005. [PMID:20643476 DOI:10.1016/j.jaci.2010.05.021]

23. **Kobayashi I,** Shiari R, Yamada M, Kawamura N, Okano M, Yara A, Iguchi A, Ishikawa N, Ariga T, Sakiyama Y, Ochs HD, Kobayashi K. Novel mutations of FOXP3 in two Japanese patients with immune dysregulation, polyendocrinopathy, enteropathy, X linked syndrome (IPEX). *J Med Genet* 2001;**38**: 874-876. [PMID:11768393 DOI:10.1136/jmg.38.12.874]

24. **Owen CJ,** Jennings CE, Imrie H, Lachaux A, Bridges NA, Cheetham TD, Pearce SH. Mutational analysis of the FOXP3 gene and evidence for genetic heterogeneity in the immunodysregulation, polyendocrinopathy, enteropathy syndrome. *J Clin Endocrinol Metab* 2003;**88**: 6034-6039. [PMID:14671208 DOI:10.1210/jc.2003-031080]

25. **Fuchizawa T,** Adachi Y, Ito Y, Higashiyama H, Kanegane H, Futatani T, Kobayashi I, Kamachi Y, Sakamoto T, Tsuge I, Tanaka H, Banham AH, Ochs HD, Miyawaki T. Developmental changes of FOXP3-expressing CD4+ CD25+ regulatory T cells and their impairment in patients with

FOXP3 gene mutations. *Clin Immunol* 2007;**125**: 237-246. [PMID:17916446 DOI:10.1016/j.clim.2007.08.004]

26. **Rubio-Cabezas O,** Minton JA, Caswell R, Shield JP, Deiss D, Sumnik Z, Cayssials A, Herr M, Loew A, Lewis V, Ellard S, Hattersley AT. Clinical heterogeneity in patients with FOXP3 mutations presenting with permanent neonatal diabetes. *Diabetes Care* 2009;**32**: 111-116. [PMID:18931102 DOI:10.2337/dc08-1188]
27. **Duclaux-Loras R,** Collardeau-Frachon S, Nancey S, Fabien N, Kaiserlian D, Lachaux A. Long-term disease course in a patient with severe neonatal IPEX syndrome. *Clin Res Hepatol Gastroenterol* 2015;**39**: e43-e47. [PMID:25977242 DOI:10.1016/j.clinre.2015.03.006]
28. **Rao A,** Kamani N, Filipovich A, Lee SM, Davies SM, Dalal J, Shenoy S. Successful bone marrow transplantation for IPEX syndrome after reduced-intensity conditioning. *Blood* 2007;**109**: 383-385. [PMID:16990602 DOI:10.1182/blood-2006-05-025072]
29. **Moudgil A,** Perriello P, Loechelt B, Przygodzki R, Fitzerald W, Kamani N. Immunodysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome: an unusual cause of proteinuria in infancy. *Pediatr Nephrol* 2007;**22**: 1799-1802. [PMID:17629750 DOI:10.1007/s00467-007-0532-0]
30. **Xavier-da-Silva MM,** Moreira-Filho CA, Suzuki E, Patricio F, Coutinho A, Carneiro-Sampaio M. Fetal-onset IPEX: report of two families and review of literature. *Clin Immunol* 2015;**156**: 131-140. [PMID:25546394 DOI:10.1016/j.clim.2014.12.007]
31. **De Benedetti F,** Insalaco A, Diamanti A, Cortis E, Muratori F, Lamioni A, Carsetti R, Cusano R, De Vito R, Perroni L, Gambarara M, Castro M, Bottazzo GF, Ugazio AG. Mechanistic associations of a mild phenotype of immunodysregulation, polyendocrinopathy, enteropathy, x-linked syndrome. *Clin Gastroenterol Hepatol* 2006;**4**: 653-659. [PMID:16630773 DOI:10.1016/j.cgh.2005.12.014]

32. **Wong T,** Li MM, Yu M, Lu CX, Zhang HB, Ping F, Zhang Q, Qi CJ, Zheng J, Wang XJ. Genetic and clinical features of 3 cases of neonatal diabetes mellitus caused by forkhead box P3 gene. *Chin J Diabetes Mellitus* 2018;**10**: 596-600. [DOI:10.3760/cma.j.issn.1674-5809.2018.09.006]

33. **Wildin R,** Smyk-Pearson S, Filipovich AH. Clinical and molecular features of the immunodysregulation, polyendocrinopathy, enteropathy, X linked (IPEX) syndrome. *J Med Genet* 2002;**39**: 537-545. [PMID:12161590 DOI:10.1136/jmg.39.8.537]

34. **Zennaro D,** Scala E, Pomponi D, Caprini E, Arcelli D, Gambineri E, Russo G, Mari A. Proteomics plus genomics approaches in primary immunodeficiency: the case of immune dysregulation, polyendocrinopathy, enteropathy, X‐linked (IPEX) syndrome. *Clin Exp Immunol* 2012;**167**: 120-128. [PMID:22132891 DOI:10.1111/j.1365-2249.2011.04492.x]

35. **Halabi‐Tawil M,** Ruemmele F, Fraitag S, Rieux‐Laucat F, Neven B, Brousse N, De Prost Y, Fischer A, Goulet O, Bodemer C. Cutaneous manifestations of immune dysregulation, polyendocrinopathy, enteropathy, X‐linked (IPEX) syndrome. *Br J Dermatol* 2009;**160**: 645-651. [PMID:18795917 DOI:10.1111/j.1365-2133.2008.08835.x]

36. **Patey-Mariaud de Serre N,** Canioni D, Ganousse S, Rieux-Laucat F, Goulet O, Ruemmele F, Brousse N. Digestive histopathological presentation of IPEX syndrome. *Mod Pathol* 2009;**22**: 95-102. [PMID:18820676 DOI:10.1038/modpathol.2008.161]

37. **Chatila TA,** Blaeser F, Ho N, Lederman HM, Voulgaropoulos C, Helms C, Bowcock AM. JM2, encoding a fork head–related protein, is mutated in X-linked autoimmunity–allergic disregulation syndrome. *J Clin Invest* 2000;**106**: R75-R81. [PMID:11120765 DOI:10.1172/JCI11679]

38. **Okou DT,** Mondal K, Faubion WA, Kobrynski LJ, Denson LA, Mulle JG, Ramachandran D, Xiong Y, Svingen P, Patel V, Bose P, Waters JP,

Prahalad S, Cutler DJ, Zwick ME, Kugathasan S. Exome sequencing identifies a novel FOXP3 mutation in a 2-generation family with inflammatory bowel disease. *J Pediatr Gastroenterol Nutr* 2014;**58**: 561-8. [PMID:24792626 DOI:10.1097/MPG.0000000000000302]

39. **Baris S,** Schulze I, Ozen A, Aydiner EK, Altuncu E, Karasu GT, Ozturk N, Lorenz M, Schwarz K, Vraetz T, Ehl S, Barian IB. Clinical heterogeneity of immunodysregulation, polyendocrinopathy, enteropathy, X-linked: pulmonary involvement as a non-classical disease manifestation. *J Clin Immunol* 2014;**34**: 601-606. [PMID:24916357 DOI:10.1007/s10875-014-0059-7]

40. **Louie RJ,** Tan QKG, Gilner JB, Rogers RC, Younge N, Wechsler SB, McDonald MT, Gordon B, Saski CA, Jones JR, Chapman SJ, Stevenson RE, Sleasman JW, Friez MJ. Novel pathogenic variants in FOXP3 in fetuses with echogenic bowel and skin desquamation identified by ultrasound. *Am J Med Genet A* 2017;**173**: 1219-1225. [PMID:28317311 DOI:10.1002/ajmg.a.38144]

41. **Chen CA,** Chung WC, Chiou YY, Yang YJ, Lin YC, Ochs HD, Shieh CC. Quantitative analysis of tissue inflammation and responses to treatment in immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome, and review of literature. *J Microbiol Immunol Infect* 2016;**49**: 775-782. [PMID:26748735 DOI:10.1016/j.jmii.2015.10.015]

42. **Park E,** Chang HJ, Shin JI, Lim BJ, Jeong HJ, Lee KB, Moon KC, Kang HG, Ha IS, Cheong HI. Familial IPEX syndrome: different glomerulopathy in two siblings. *Pediatr Int* 2015;**57**: e59-e61. [PMID:25712815 DOI:10.1111/ped.12570]

43. **Lopes JE,** Torgerson TR, Schubert LA, Anover SD, Ocheltree EL, Ochs HD, Ziegler SF. Analysis of FOXP3 reveals multiple domains required for its function as a transcriptional repressor. *J Immunol* 2006;**177**: 3133-3142. [PMID:16920951 DOI:10.4049/jimmunol.177.5.3133]

44. **Hashimura Y,** Nozu K, Kanegane H, Miyawaki T, Hayakawa A, Yoshikawa N, Nakanishi K, Takemoto M, Iijima K, Matsuo M. Minimal change nephrotic syndrome associated with immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. *Pediatr Nephrol* 2009;**24**: 1181-1186. [PMID:19189134 DOI:10.1007/s00467-009-1119-8]

45. **Li B,** Samanta A, Song X, Iacono KT, Brennan P, Chatila TA, Roncador G, Banham AH, Riley JL, Wang Q, Shen Y, Saouaf SJ, Greene MI. FOXP3 is a homo-oligomer and a component of a supramolecular regulatory complex disabled in the human XLAAD/IPEX autoimmune disease. *Int Immunol* 2007;**19**: 825-835. [PMID:17586580 DOI:10.1093/intimm/dxm043]

46. **Luo Y,** Chen J, Fang Y, Lou J, Yu J. A case of Metaplastic atrophic gastritis in immune Dysregulation, Polyendocrinopathy, Enteropathy, X-linked (IPEX) syndrome. *BMC Pediatr* 2018;**18**: 191. [PMID:29907148 DOI:10.1186/s12887-018-1169-9]

47. **Dong WF,** Cao Q, Wang J, LI BR. Two case reports about immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome and literature review. *Chin Pediatr Emerg Med* 2018;**25**: 753-756. [DOI:10.3760/cma.j.issn.1673-4912.2018.10.009]

48. **Rodrigo R,** Atapattu N, de Silva KS. IPEX syndrome with membrano-proliferative nephrotic syndrome. *Ceylon Med J* 2013;**58**. [PMID:23549728 DOI:10.4038/cmj.v58i1.5368]

49. **Nademi Z,** Slatter M, Gambineri E, Mannurita S, Barge D, Hodges S, Bunn S, Thomas J, Haugk B, Hambleton S, Flood T, Cant A, Abinun M, Gennery A. Single centre experience of haematopoietic SCT for patients with immunodysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. *Bone Marrow Transplant* 2014;**49**: 310-312. [PMID:24270390 DOI:10.1038/bmt.2013.181]

50. **Harbuz R,** Lespinasse J, Boulet S, Francannet C, Creveaux I, Benkhelifa M, Jouk PS, Lunardi J, Ray PF. Identification of new FOXP3 mutations

and prenatal diagnosis of IPEX syndrome. *Prenat Diagn* 2010;**30**: 1072-1078. [PMID:20842625 DOI:10.1002/pd.2613]

51. **Shehab O,** Tester DJ, Ackerman NC, Cowchock FS, Ackerman MJ. Whole genome sequencing identifies etiology of recurrent male intrauterine fetal death. *Prenat Diagn* 2017;**37**: 1040-1045. [PMID:28833278 DOI:10.1002/pd.5142]
52. **Zhu Q,** Yuan K, Wang C, Fang Y, Zhu J, Liang L. Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome in two cases. *Chin J Med Genet* 2018;**35**: 389-392. [PMID:29896738 DOI:10.3760/cma.j.issn.1003-9406.2018.03.018]
53. **Bindl L,** Torgerson T, Perroni L, Youssef N, Ochs HD, Goulet O, Ruemmele FM. Successful use of the new immune-suppressor sirolimus in IPEX (immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome). *J Pediatr* 2005;**147**: 256-259. [PMID:16126062 DOI:10.1016/j.jpeds.2005.04.017]
54. **An YF,** Xu F, Wang M, Zhang ZY, Zhao XD. Clinical and Molecular Characteristics of Immunodysregulation, Polyendocrinopathy, Enteropathy, X‐Linked Syndrome in China. *Scand J Immunol* 2011;**74**: 304-309. [PMID:21595732 DOI:10.1111/j.1365-3083.2011.02574.x]
55. **McMurchy AN,** Gillies J, Allan SE, Passerini L, Gambineri E, Roncarolo MG, Bacchetta R, Levings MK. Point mutants of forkhead box P3 that cause immune dysregulation, polyendocrinopathy, enteropathy, X-linked have diverse abilities to reprogram T cells into regulatory T cells. *J Allergy Clin Immunol* 2010;**126**: 1242-1251. [PMID:21036387 DOI:10.1016/j.jaci.2010.09.001]
56. **Reichert SL,** McKay EM, Moldenhauer JS. Identification of a novel nonsense mutation in the FOXP3 gene in a fetus with hydrops—Expanding the phenotype of IPEX syndrome. *Am J Med Genet A* 2016;**170**: 226-232. [PMID:26395338 DOI:10.1002/ajmg.a.37401]

57. **Rae W,** Gao Y, Bunyan D, Holden S, Gilmour K, Patel S, Wellesley D, Williams A. A novel FOXP3 mutation causing fetal akinesia and recurrent male miscarriages. *Clin Immunol* 2015;**161**: 284-285. [PMID:26387632 DOI:10.1016/j.clim.2015.09.006]

58. **Savova R,** Arshinkova M, Houghton J, Konstantinova M, Gaydarova M, Georgieva E, Popova G, Genova V, Tomova N, Anadoliiska A, Koprivarova K. Clinical case of immune dysregulation, polyendocrinopaty, enteropathy, X-linked (IPEX) syndrome with severe immune deficiency and late onset of endocrinopathy and enteropathy. *Case Rep Med* 2014;**2014**. [PMID:24982679 DOI:10.1155/2014/564926]

59. **Tuijnenburg P,** Cuadrado E, Bosch AM, Kindermann A, Jansen MH, Alders M, van Leeuwen EM, Kuijpers TW. Humoral Immunodeficiency with hypotonia, feeding difficulties, enteropathy, and mild eczema caused by a classical FOXP3 mutation. *Front Pediatr* 2017;**5**: 37. [PMID:28289675 DOI:10.3389/fped.2017.00037]

60. **Hoshino A,** Kanegane H, Nishi M, Tsuge I, Tokuda K, Kobayashi I, Imai K, Morio T, Takagi M. Identification of autoantibodies using human proteome microarrays in patients with IPEX syndrome. *Clin Immunol* 2019;**203**: 9-13. [PMID:30951839 DOI:10.1016/j.clim.2019.03.011]

61. **Agakidis C,** Agakidou E, Sarafidis K, Papoulidis I, Xinias I, Farmaki E. Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome associated with a novel mutation of FOXP3 gene. *Front Pediatr* 2019;**7**: 20. [PMID:30805323 DOI:10.3389/fped.2019.00020]

62. **Vasiljevic A,** Poreau B, Bouvier R, Lachaux A, Arnoult C, Fauré J, Cordier MP, Ray PF. Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome and recurrent intrauterine fetal death. *Lancet* 2015;**385**: 2120. [PMID:26009232 DOI:10.1016/S0140-6736(15)60773-5]

63. **Scaillon M,** Van Biervliet S, Bontems P, Dorchy H, Hanssens L, Ferster A, Segers V, Cadranel S. Severe gastritis in an insulin-dependent child with an IPEX syndrome. *J Pediatr Gastroenterol Nutr* 2009;**49**: 368-370. [PMID:19633572 DOI:10.1097/MPG.0b013e3181a159de]

64. **Seidel MG,** Boztug K, Haas OA. Immune dysregulation syndromes (IPEX, CD27 deficiency, and others): always doomed from the start? *J Clin Immunol* 2016;**36**: 6-7. [PMID:26661331 DOI:10.1007/s10875-015-0218-5]

65. **Yamauchi T,** Takasawa K, Kamiya T, Kirino S, Gau M, Inoue K, Hoshino A, Kashimada K, Kanegane H, Morio T. Hematopoietic stem cell transplantation recovers insulin deficiency in type 1 diabetes mellitus associated with IPEX syndrome. *Pediatr Diabetes* 2019;**20**: 1035-1040. [PMID:31322807 DOI:10.1111/pedi.12895]

66. **Costa‐Carvalho BT,** De Moraes‐Pinto MI, De Almeida L, de Seixas Alves M, Maia R, de Souza RL, Barreto M, Lourenço L, Vicente AM, Coutinho A, Carneiro-Sampaio M. A remarkable depletion of both naive CD4+ and CD8+ with high proportion of memory T cells in an IPEX infant with a FOXP3 mutation in the forkhead domain. *Scand J Immunol* 2008;**68**: 85-91. [PMID:18489537 DOI:10.1111/j.1365-3083.2008.02055.x]

67. **Suzuki S,** Makita Y, Mukai T, Matsuo K, Ueda O, Fujieda K. Molecular basis of neonatal diabetes in Japanese patients. *J Clin Endocrinol Metab* 2007;**92**: 3979-3985. [PMID:17635943 DOI:10.1210/jc.2007-0486]

68. **Colobran R,** Alvarez de la Campa E, Soler-Palacín P, Martín-Nalda A, Pujol-Borrell R, de la Cruz X, Martínez-Gallo M. Clinical and structural impact of mutations affecting the residue Phe367 of FOXP3 in patients with IPEX syndrome. *Clin Immunol* 2016;**163**: 60-65. [PMID:26748374 DOI:10.1016/j.clim.2015.12.014]

69. **Wildin RS,** Ramsdell F, Peake J, Faravelli F, Casanova J-L, Buist N, Levy-Lahad E, Mazzella M, Goulet O, Perroni L, Bricarelli FD, Byrne G,

McEuen M, Proll S, Appleby M, Brunkow ME. X-linked neonatal diabetes mellitus, enteropathy and endocrinopathy syndrome is the human equivalent of mouse scurfy. *Nat Genet* 2001;**27**: 18-20. [PMID:11137992 DOI:10.1038/83707]

70. **Baud O,** Goulet O, Canioni D, Le Deist F, Radford I, Rieu D, Dupuis-Girod S, Cerf-Bensussan N, Cavazzana-Calvo M, Brousse N, Fischer A, Casanova JL. Treatment of the immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome (IPEX) by allogeneic bone marrow transplantation. *N Engl J Med* 2001;**344**: 1758-1762. [PMID:11396442 DOI:10.1056/NEJM200106073442304]
71. **Tanaka H,** Tsugawa K, Kudo M, Sugimoto K, Kobayashi I, Ito E. Low-dose cyclosporine A in a patient with X-linked immune dysregulation, polyendocrinopathy and enteropathy. *Eur J Pediatr* 2005;**164**: 779-780. [PMID:16091915 DOI:10.1007/s00431-005-1746-4]
72. **Zhan H,** Sinclair J, Adams S, Cale CM, Murch S, Perroni L, Davies G, Amrolia P, Qasim W. Immune reconstitution and recovery of FOXP3 (forkhead box P3)-expressing T cells after transplantation for IPEX (immune dysregulation, polyendocrinopathy, enteropathy, X-linked) syndrome. *Pediatrics* 2008;**121**: e998-e1002. [PMID:18316354 DOI:10.1542/peds.2007-1863]
73. **Bennett CL,** Christie J, Ramsdell F, Brunkow ME, Ferguson PJ, Whitesell L, Kelly TE, Saulsbury FT, Chance PF, Ochs HD. The immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome (IPEX) is caused by mutations of FOXP3. *Nat Genet* 2001;**27**: 20-21. [PMID:11137993 DOI:10.1038/83713]
74. **Nieves DS,** Phipps RP, Pollock SJ, Ochs HD, Zhu Q, Scott GA, Ryan CK, Kobayashi I, Rossi TM, Goldsmith LA. Dermatologic and immunologic findings in the immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. *Arch Dermatol* 2004;**140**: 466-472. [PMID:15096376 DOI:10.1001/archderm.140.4.466]

75. **Taddio A,** Faleschini E, Valencic E, Granzotto M, Tommasini A, Lepore L, Andolina M, Barbi E, Ventura A. Medium-term survival without haematopoietic stem cell transplantation in a case of IPEX: insights into nutritional and immunosuppressive therapy. *Eur J Pediatr* 2007;**166**: 1195-1197. [PMID:17205241 DOI:10.1007/s00431-006-0395-6]

76. **Kasow KA,** Morales-Tirado VM, Wichlan D, Shurtleff SA, Abraham A, Persons DA, Riberdy JM. Therapeutic in vivo selection of thymic-derived natural T regulatory cells following non-myeloablative hematopoietic stem cell transplant for IPEX. *Clin Immunol* 2011;**141**: 169-176.]PMID:21865090 DOI:10.1016/j.clim.2011.07.005]

77. **He TY,** Xia Y, Liang FF, Luo Y, Jia SL, Yang J. IPEX with congenital ichthyosiform skin lesions: a case report and literature review. *J Clin Pediatr* 2017;**35**: 620-624. [DOI:10.3969/j.issn.1000-3606.2017.08.015]

78. **Griswold C,** Durica AR, Dennis LG, Jewell AF. Prenatal bowel findings in male siblings with a confirmed FOXP3 mutation. *J Ultrasound Med* 2018;**37**: 1033-1037. [PMID:28960390 DOI:10.1002/jum.14428]

79. **Bachelerie M,** Merlin E, Beltzung F, Franck F, Joubert J, Hall T, Fraitag S, D'Incan M. Érythrodermie révélatrice d'un syndrome IPEX. *Elsevier Masson 2019;***146**: 807-811. [DOI:10.1016/j.annder.2019.04.026]

80. **Levy-Lahad E,** Wildin RS. Neonatal diabetes mellitus, enteropathy, thrombocytopenia, and endocrinopathy: further evidence for an X-linked lethal syndrome. *J Pediatr* 2001;**138**: 577-580. [PMID:11295725 DOI:10.1067/mpd.2001.111502]

81. **Banerjee‐Basu S,** Baxevanis AD. Structural analysis of disease‐causing mutations in the P‐subfamily of forkhead transcription factors. *Proteins* 2004;**54**: 639-647. [PMID:14997560 DOI:10.1002/prot.10621]

82. **Barzaghi F,** Hernandez LCA, Neven B, Ricci S, Kucuk ZY, Bleesing JJ, Nademi Z, Slatter MA, Ulloa ER, Shcherbina A, Roppelt A, Worth A, Silva J, Aiuti A, Murguia-Favela L, Speckmann C, Carneiro-Sampaio M,

Fernandes JF, Baris S, Baris S, Ozen A, Karakoc-Aydiner E. Long-term follow-up of IPEX syndrome patients after different therapeutic strategies: an international multicenter retrospective study. *J Allergy Clin Immunol* 2018;**141**: 1036-1049. e5. [PMID:29241729 DOI:10.1016/j.jaci.2017.10.041]

83. **Ge T,** Wang Y, Che Y, Xiao Y, Zhang T. Atypical late-onset immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome with intractable diarrhea: a case report. *Front Pediatr* 2017;**5**: 267. [PMID:29312905 DOI:10.3389/fped.2017.00267]
84. **Smith E,** Greeley SAW, Ye H, Torgerson TR, Dimmitt R, Atkinson P, Philips J, Goldman F. Extremely early onset IPEX syndrome caused by a novel small exonic deletion in FOXP3. *J Pediatr Gastroenterol Nutr* 2016;**63**: e119. [PMID:25187107 DOI:10.1097/MPG.0000000000000554]
85. **Alkorta-Aranburu G,** Carmody D, Cheng Y, Nelakuditi V, Ma L, Dickens JT, Das S, Greeley S, Del Gaudio D. Phenotypic heterogeneity in monogenic diabetes: the clinical and diagnostic utility of a gene panel-based next-generation sequencing approach. *Mol Genet Metab* 2014;**113**: 315-320. [PMID:25306193 DOI:10.1016/j.ymgme.2014.09.007]
86. **Burroughs LM,** Storb R, Leisenring WM, Pulsipher MA, Loken MR, Torgerson TR, Ochs HD, Woolfrey AE. Intensive postgrafting immune suppression combined with nonmyeloablative conditioning for transplantation of HLA-identical hematopoietic cell grafts: results of a pilot study for treatment of primary immunodeficiency disorders. *Bone Marrow Transplant* 2007;**40**: 633-642. [PMID:17660844 DOI:10.1038/sj.bmt.1705778]
87. **Peake JE,** McCrossin RB, Byrne G, Shepherd R. X-linked immune dysregulation, neonatal insulin dependent diabetes, and intractable diarrhoea. *Arch Dis Child Fetal Neonatal Ed* 1996;**74**: F195-F199. [PMID:8777684 DOI:10.1136/fn.74.3.f195]

88. **Bennett CL,** Brunkow ME, Ramsdell F, O'Briant KC, Zhu Q, Fuleihan RL, Shigeoka AO, Ochs HD, Chance PF. A rare polyadenylation signal mutation of the FOXP3 gene (AAUAAA→ AAUGAA) leads to the IPEX syndrome. *Immunogenetics* 2001;**53**: 435-439. [PMID:11685453 DOI:10.1007/s002510100358]

89. **Dorsey MJ,** Petrovic A, Morrow MR, Dishaw LJ, Sleasman JW. FOXP3 expression following bone marrow transplantation for IPEX syndrome after reduced-intensity conditioning. *Immunol Res* 2009;**44**: 179-184. [PMID:19471859 DOI:10.1007/s12026-009-8112-y]